Commentary
Volume 13:1, 2022

Journal of Molecular Biomarkers & Diagnosis

ISSN: 2155-9929 Open Access

Insights on Cytogenetic Analysis

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Commentary

The study of normal chromosomes and chromosome aberrations is the focus of cytogenetics, a branch of pathology and genetics. Classical cytogenetics allows for the microscopic examination of entire chromosomes in order to determine their number and structure. To evaluate submicroscopic chromosomal regions, molecular cytogenetics employs specialised techniques such as Fluorescence *In Situ* Hybridization (FISH) and array comparative genomic hybridization (aCGH). To investigate constitutional and acquired chromosome abnormalities, both classical and molecular cytogenetic techniques are used. Next-generation sequencing has recently been added to the cytogenetic lab's arsenal, allowing for the precise identification of breakpoints in chromosome rearrangements. Each of these techniques will be discussed in greater detail in the following sections.

Molecular and cytogenetic diagnostics are critical components of genetic counselling. As our understanding of the human genome has grown, so has the development of efficient, accurate testing methods. The list below provides an overview of the most commonly used diagnostic tools. When the cell is in prometaphase or metaphase and the chromosomes are condensed, chromosomal analysis, or karyotyping, is best performed. A Giemsa stain, the most common staining technique, produces a distinct pattern of alternating light and dark bands that is dependent on the composition of the underlying DNA sequence. Each chromosome has its own pattern of banding. The analysis of this banding pattern can reveal structural flaws.

The disadvantage of this technique is that its sensitivity is limited, which means that minor structural abnormalities or mutations would go undetected. The benefit of this technique is that it allows you to see the entire genome at once. Cytogenetics is a branch of biology that studies chromosomes and their abnormalities. Chromosome analysis, also known as karyotyping, is the process of pairing homologous chromosomes.Chromosome abnormalities cause a variety of genetic disorders, including developmental delay, congenital malformations, mental retardation, and infertility. The use of cytogenetic analysis in the diagnosis of oncologic and hematologic disorders is critical. It aids in disease diagnosis and classification, as well as treatment regimen planning and disease monitoring.

A critical role in the diagnosis, classification, prognosis, and management of acute myeloid leukemia is cytogenetic analysis (AML). It has evolved into a vital technique for assisting doctors in identifying leukemia and providing treatment recommendations. Cytogenetic testing is typically performed on AML patients' bone marrow and aids in characterizing the aggressiveness of the leukemia. It also aids in determining treatment response and overall prognosis. In the case of AML patients, genetic testing for FLT3, CEBPA, and NPM1 mutations is now standard practice. The underlying cytogenetic abnormalities, such as chromosomal duplications, deletions, or substitutions, as well as under- or over-expression of certain proteins, are used to identify AML risk factors.

This aids in the detection of structural or numerical chromosome abnormalities. Chromosome analyses necessitate cell cultures and include chromosome harvesting, chromosome banding, microscopic analysis, and the generation of karyotypes. This type of testing can detect genomic copy number changes or minor genetic imbalances, such as chromosomal gain or loss, that traditional FISH or cytogenetic analysis methods cannot detect. It also aids in the identification of specific genes involved in a chromosomal abnormality detected using traditional FISH or cytogenetic testing methods [1-5].

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How to cite this article: Swapna, Sanike. "Insights on Cytogenetic Analysis." J Mol Biomark Diagn 13 (2022):508.

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Received 03-Jan-2022, Manuscript No. Jmbd-22-56409; **Editor assigned:** 05-Jan-2022, Pre QC No. P-56409; **Reviewed:** 17-Jan-2022, QC No.Q-56409; **Revised:** 22-Jan-2022, Manuscript No.R-56409; **Published:** 29-Jan-2022, DOI: 10.37421/ jmbd.2022.11.508.